

Newborn Screening Update

Michigan Newborn Screening Program

Fall 2007

Volume 6 Issue 3



Announcements...

Our Newborn Screening Educator, Midge McCaustland, has retired. Many thanks and appreciation for all her fine work with the newborn screening program. We miss Midge but hope she thoroughly enjoys herself in retirement.

For now please email the Newborn Screening Program at: mdch-newbornscreening@michigan.gov with any issues you would have contacted Midge about or call our main line at 517-335-9205 and you will be directed to the person who can help you out. Thank you in advance for your cooperation as we work towards getting this position filled.

Newborn Screening for Cystic Fibrosis Implemented October 1, 2007

Follow-up for presumptive positive cystic fibrosis results will be handled by The Cystic Fibrosis Newborn Screening Coordinating Center at the University of Michigan Health System under the direction of Dr. Samya Nasr. The center will notify physicians and parents of the presumptive positive newborn screen and will coordinate with the designated accredited Cystic Fibrosis Care Centers who will arrange the necessary sweat chloride testing.

The Cystic Fibrosis Newborn Screening Coordinating Center phone number is 734-647-8938; fax 734-936-7918.

Please see page 7 for announcement of Cystic Fibrosis Newborn Screening testing which includes list of the accredited Michigan CF Centers. Page 8 has the Cystic Fibrosis Fact Sheet.

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Contacts

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NBS Coordinator
Vacant Position

NBS Educator
Vacant Position

Laboratory Manager
Harry Hawkins
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NBS Accountant
Valerie Klasko
(517) 241-5583

Updates and Reminders ...

Michigan Department of Community Health Newborn Screening NICU

Coordinators Open House...

was held September 27, 2007 in the newborn screening follow-up offices in downtown Lansing. The day started out with an overview of the program and lots of good discussion. After lunch there was a trip across town for a lab tour.

Michigan Newborn Screening Program — A First Step to Your Baby's Health brochures:

The updated English brochures are now available. The brochures are shipped in quantities of 50 per package at no charge. Contact Valerie Klasko to order 517-241-5583 or KLASKO@michigan.gov

Improved Turn Around Time with Couriers and Faxes

The Newborn Screening Program is working to improve turn around time on both the delivery and reporting components of the process. Arrangements have been made to have Quest Diagnostics and United Parcel Service bring samples to the laboratory overnight and in time to be tested the next day. New hospitals have been systematically added and now more than sixty percent of the samples arrive this way. However, sometimes a single envelope contains samples collected over several days. This indicates that specimen "batching" is still a problem in some hospitals and causes unnecessary delays in testing.

The laboratory also has been faxing newborn screening results to hospitals since January 2007 and gradually adding hospitals to the list of fax recipients. The fax option has replaced U.S. mail delivery for forty percent of the reports sent. Please call Harry Hawkins (517) 335-8095 if your hospital is interested in greatly improved turn around time.

Parental Refusal Forms:

If a parent should refuse the newborn screening could you please fax us a copy of the signed Parental Refusal Form your institution uses for our records (fax 517-335-9419). We often find out about missed cases from Newborn Hearing when they receive a hearing slip but find no matching blood specimen in the database. If we had the refusal forms on file we would know not to contact your institution about getting a family in for the missed newborn screening blood test.

Quarterly Report Cards and Newborn Screening Newsletter:

Just a reminder that when the hospital coordinator positions were established in each birthing hospital one of the tasks the coordinators were asked to take on was disseminating the quarterly report cards and newborn screening newsletters to the appropriate personal within their hospitals so we would only have to do one mailing per hospital birthing unit. We were hoping this would simplify the process for everyone involved but if it's not working for you we definitely want to know. We have heard from some nurses and lab personnel that they do not get one or the other of these documents and on these occasions we contact the coordinator to let them know. Please drop us a line at mdch-newbornscreening@michigan.gov and let us know if you are having problems.

Returning the blue initial NBS specimen card for credit:

Please include the completed NBS Card Replacement Form with the top copy of the blue initial NBS specimen card when requesting credit. Please send both to the address noted on the form.

Changes in the First and Repeat newborn screening cards

The new first sample and repeat sample collection kits are in stock. There are some changed / added fields. An email was sent to the hospital coordinators in July to address this and is reproduced here on pages 9 to 11.

A price increase for the cards went into effect October 1, 2007. A notification letter of the price increase went out to hospital administrators August 20, 2007. The new price is \$85.61 per card and \$2,140.25 per package of 25 cards.

This latest printing of the newborn screening cards has a pink 'instruction' booklet in each package. Per lab manager, Harry Hawkins, these instructions are not to be used. Use instructions on back of the card.

We have received some questions and would like to share those with you in an effort to help everyone:

- Q1. Can we finish up the current packs of old cards we have or should we be trading them in for the new cards? Is there a "drop dead" date by which all hospitals must be using the new cards only?
- A. There is no "drop dead" date for using the new cards only. Please trade in the old cards for new ones as soon as possible.
- Q2. The question regarding "antibiotics" - does this include intrapartum antibiotics given to the mom since they do pass into the baby's blood stream prior to delivery or are you referring only to post delivery antibiotics administered to the baby?
- A. Yes, includes intrapartum antibiotics.
- Q3. For the Repeat Screening card, we instruct the parents to return to the lab for this repeat draw before the baby is "2 weeks old" - is this still correct?
- A. For a repeat screen, needed on follow-up to a first sample, the general answer to this is yes. However, newborn screening follow-up sends out a letter to the physician of record, listed on the card, with instructions on when to obtain the repeat newborn screen. If the baby is not in the NICU, a letter is also sent to the parent informing them a repeat screen is needed. If blood transfusions and TPN have not been given to the baby we request the repeat ASAP to resolve any borderline positive or inconclusive test results.
- Q4. If the baby spent a brief period (less than 24 hours) in the NICU but was in the Newborn Nursery at the time that the screen was drawn, should we still put "yes" under NICU/ Special Care?

A. Completely shade in oval “no” or “yes” to indicate if the newborn was in an NICU or special care nursery **when** the specimen was collected. If the baby is not in the NICU at the time the specimen is collected, shade NICU oval No.

Q5. The rest of the rules of filling out the other blanks still apply, correct?

Ethnicity — check either Hispanic or Non-Hispanic

Race — check only one bubble

Use military time

Use grams for weight

Etc, etc, etc...

A. Yes. Just a reminder, it is very important to fill in either the Hispanic or Non-Hispanic box **and** in addition fill in one of the six boxes for race. Both are necessary to identify certain populations of interest or at risk of particular conditions. Examples:

1. If a baby has at least one parent who identifies as Hispanic and both parents are Black, the card should be marked Hispanic and Black.
2. If one parent is Black and the other is White and one parent identifies as Hispanic, the card should be marked Hispanic and Multi-Racial.
3. If one parent is Black and one parent is White and neither parent identifies as Hispanic, the card should be marked Non-Hispanic and Multi-Racial.

Q6. If the mom is treated with betamethasone during her pregnancy, do we include this? What might be some common steroids we should be aware of?

A. Yes. Betamethasone, dexamethasone, prednisone.

Q7. Regarding filling out the “race” box, Previous instructions stated, “If the infant is of mixed race and has one ‘white’ parent, select the race of the NON-WHITE parent. If the infant is of mixed race and both parents are non-white, select the ‘MULTI-RACIAL’ bubble.” Is this still correct?

No. See Q5 above. We will correct our guidelines.

Q8. Will the fax number be a required field?

A. Yes

Q9. If the baby will be adopted, should we put the birth mothers information in the boxes and then write the adoptive parents contact info in the margin?

A. If the newborn is going to be released at birth to adoptive or foster parents, provide contact information of adoptive or foster mother. Please note, in black ink above mothers name, that contact information is for adoptive or foster mother. Do not place sticky notes on the card or use red ink, neither will be recorded when the card is scanned into the system. If contact information on new parents, foster parents, or the adoption agency is not on the card, we will not be able to contact the family if necessary. We would like to avoid calling the birth mother if she is no longer responsible for the care of the newborn.

The following story was originally published in: "About Children's" magazine Winter 2006, a publication of the Children's Hospital of Michigan. Thank you for letting us share Laine's story in our newsletter.

Early Detection and Treatment Saves Young Girl's Life

BY MICHAEL HODGES



Laine Decker was six days old when her parents, Jennifer and Jeffrey, got the phone call you never want to get. Their pediatrician in Battle Creek said she needed to see Laine – immediately.

Tests taken at birth showed possible abnormalities. Could Jeffrey and Jennifer bring her in right away?

"We were terrified," says Jennifer, "especially given that when I asked our pediatrician whether Laine could die from this, she had no answer, because she'd never heard of the condition."

The condition is Very Long-Chain Acyl-CoA Dehydrogenase Deficiency, or VLCADD, an extremely rare metabolic disorder where the body cannot break down fatty acids because of a missing, or malfunctioning, enzyme.

Although their doctor said she was pretty sure this was a false positive, the Deckers were told in no uncertain terms to get to Children's Hospital of Michigan, the only state-designated medical institution in Michigan with a follow-up and treatment program for infants born with a positive newborn screen indicative of an inborn error of metabolism.

Praying that it was all just an awful mistake, Jennifer, Jeffrey and Laine made the two-hour drive to Detroit for further tests. Given the circumstances, it was a drive that seemed to have no end.

And the truth is — Jennifer had a premonition.

"Maybe it was just typical mother's worry," she says, "but from the day we brought her home, I kept asking my husband whether everything was okay with Laine. Something just didn't seem right."

"While we waited for test results, we had to treat Laine like she had VLCADD," says Jennifer. Not having any answers just made me feel like I couldn't relax at all."

The final diagnosis? Laine has a "mild" form of the deficiency, caused by a recessive gene, that is unlikely to cause any major medical problems in Laine.

Laine Decker is a happy toddler thanks to early detection of a rare metabolic disorder.



“Our experience has been awesome. The staff is incredibly supportive and caring. That’s part of why we feel so grateful.”

Jennifer Decker

VLCADD is one of 48 conditions that Michigan tests all newborns for within 36 hours of birth. The test has the potential to save lives.

The consequences of not identifying VLCADD can be dire. Children with VLCADD might toddle along just fine — until a cold or flu keeps them from eating. Unable to break down fats while “fasting,” children with VLCADD end up with hypoglycemia, breathing problems, seizure, coma and possible death,” says Jennifer, who’s a nurse.

When a child with VLCADD stops eating, they have to be hospitalized so a dextrose-rich fluid can be pumped into them. Laine’s been relatively fortunate. At 14 months now, she’s only had to be hospitalized once in Battle Creek because of an intestinal flu.

Jennifer estimates the family has made five trips over the past year to Children’s, but that doesn’t count calls to check on lab results and get advice.

Staff at Children’s metabolic disorders clinic, says Jennifer, have been remarkable.

“Our experience has been awesome,” she says. “The staff is incredibly supportive and caring. That’s part of why we feel so grateful.”

In particular, she says, certified genetic counselor Peggy Rush has been a lifeline — a calm, reassuring voice that Jennifer can always rely on to calm her down.

“She’s just gone above and beyond,” Jennifer says, “in helping us learn about Laine’s condition. I don’t know what to say. We’ve written letters to the hospital CEO letting him know about her professionalism and patience.”

Because of their positive experience with Children’s, the Deckers are launching a fundraiser to raise money for research on VLCADD, which they will donate to Children’s Hospital. Already, she says, the family has sent out 150 letters to friends and relatives who are concerned about their little girl, asking that they consider a contribution.

Still, the reality is you wouldn’t suspect a thing if you met Laine, a feisty little girl with blue eyes and strawberry-blonde hair who loves scribbling with chalk on the sidewalk, and tormenting her older sister, Paige, who’s 5, by stealing her Barbie.

“Laine will always have to be hospitalized as a precaution anytime she can’t eat,” says Jennifer. “But, we’re doing well. I think it took a good year to come to grips with having a chronic issue.”

In virtually every other respect, Laine is just your typical little bundle of energy. “We’re lucky,” Jennifer says. “Laine has a pretty normal life. We know we’re blessed.”



STATE OF MICHIGAN

DEPARTMENT OF COMMUNITY HEALTH
LANSING

JENNIFER M. GRANHOLM
GOVERNOR

JANET OLSZEWSKI
DIRECTOR

September 2007

**Newborn Screening for Cystic Fibrosis
Implementation: October 1, 2007**

Dear Practitioner:

We would like to inform you that beginning October 1, 2007, cystic fibrosis will be added to the current dried blood spot screening panel of 48 disorders.

Cystic fibrosis (CF) is one of the most common autosomal recessive genetic disorders affecting children. It occurs in about 1 in 3,500 white newborn infants, with a lower incidence in other ethnic groups. Although a specific cure for CF is still needed, treatment at specialized Cystic Fibrosis Centers has greatly improved the quality of life for people with CF. The average life expectancy has also increased significantly from less than ten years, when the disease was first identified over 40 years ago, to the current mean above the mid-thirties.

The objective of newborn screening for CF is to identify most affected children and make early referral to a specialty center accredited by the national Cystic Fibrosis Foundation that offers aggressive intervention to allow normal growth and delay the onset of lung disease. Any delay can result in irreversible bronchiectatic changes with dramatic impact on the lungs' vital functions.

The IRT concentration will be used by the Michigan Department of Community Health (MDCH), Newborn Screening Laboratory as the first tier in screening. It will be followed by DNA mutation analysis using a panel of 40 mutations as a second tier screen for infants with an abnormal IRT. Both tiers of the screening test for CF can be run on the dried filter paper blood spots collected as part of current newborn screening procedures. Specimen collection will not change with the addition of CF, i.e. additional samples will not be needed.

We anticipate that each year about 350 Michigan newborns will have a positive CF screen. Approximately **1 in 10** of these infants will have a diagnosis of CF. The majority of the others will be carriers of CF and will not have symptoms of the disease. Infants with a positive newborn screen will require confirmatory sweat chloride testing at a Cystic Fibrosis Center. More extensive genetic testing may be needed for a small percentage of patients. Genetic counselors will be available through the CF Care Centers for patients undergoing confirmatory testing, as well as for those identified as carriers. For children with a confirmed diagnosis of CF, ongoing assessment and treatment at a CF center are strongly recommended.

There are currently five accredited CF centers in Michigan:

- University of Michigan (Ann Arbor), serves as the state's CF NBS Coordinating Center
Phone: (734) 764-4123
- Children's Hospital of Michigan (Detroit)
Phone: (313) 745-5541
- DeVos Children's Hospital (Grand Rapids)
Phone: (616) 391-2125
- MSU Kalamazoo Center for Medical Studies (Kalamazoo) *Phone: (269) 337-6433*
- Michigan State University Cystic Fibrosis Center (Lansing) *Phone: (517) 364-5440*

Prompt communication and interpretation of results with the patient's primary care physician will be provided by MDCH and CF Coordinating Center staff. If you have any questions or concerns, please do not hesitate to contact us at (517) 335-9205 or by e-mail at mdch-newbornscreening@michigan.gov.

Sincerely,

William Young, PhD
Newborn Screening Follow Up Program



What is Cystic Fibrosis (CF)?

Cystic fibrosis is an inherited disorder that causes the body to make thick, sticky mucus. The mucus clogs the airways of the lungs and causes problems with the way the pancreas helps break down food. CF occurs in about 1 in 3,200 Caucasian newborns. It occurs less often in other ethnic and racial groups. Early diagnosis and treatment will allow children with CF the opportunity to lead longer and healthier lives.

Michigan Resources & Support

NBS & Coordinating Program for CF

University of Michigan
Phone: 734-647-8938

Children's Special Health Care Services

www.michigan.gov/cshcs
Family Phone Line
Toll-free: 1-800-359-3722

Early On[®] Michigan

www.1800earlyon.org
Toll-free: 1-800-EARLY ON

Michigan Genetics Connection

www.migeneticsconnection.org
E-mail: genetics@michigan.gov
Phone: 1-866-852-1247

Michigan Newborn Screening

Follow-up Coordinator
E-mail: MDCH-newbornscreening@michigan.gov
Toll-free: 1-866-673-9939

National Resources & Support

Cystic Fibrosis Foundation

www.cff.org
Toll-free: 1-800-344-4823

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.genetests.org

Genetic Alliance

www.geneticalliance.org

STAR-G Project

www.newbornscreening.info/Parents/facts.html

How may CF affect my child?

Symptoms in the newborn

Many newborns with CF will have no signs or symptoms of the disease at birth. Some will be born with a problem called meconium ileus where thick stool blocks the intestines. Babies with CF may also have "failure to thrive" which means they have trouble gaining weight and growing after birth.

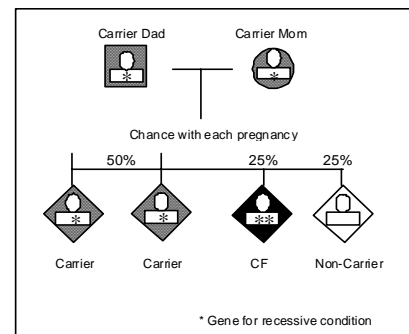
Symptoms in the older child and adult

Many children with CF will develop symptoms within the first years of life. Symptoms of CF vary, but may include:

- Coughing
- Wheezing
- Lung infections
- Sinus problems
- Poor weight gain
- Greasy stools

How does CF occur?

CF is a genetic disorder. Parents of an affected child each carry one abnormal copy of the CF gene. Carrier parents do not have symptoms of CF. In order to have CF, a person must have two abnormal copies of the CF gene. There is a 1 in 4 chance that each child will have CF when both parents carry a gene for the disorder.



How is CF treated?

CF cannot be cured, but many symptoms can be treated. Many newborns with meconium ileus will need surgery shortly after birth. Most children and adults with CF will need to eat a healthy, high-calorie diet and take vitamins to help them grow. Some people may also need medications to help them get more nutrients from the food they eat. To breathe better, many people with CF need help clearing mucus from their lungs each day. Some medications can also prevent infections and help with breathing. The Newborn Screening Program recommends that all babies with CF be treated in an accredited CF Center. Children with CF and their families also benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

**For more information please contact the Newborn Screening Program
toll-free at 1-866-673-9939 or e-mail NBS-Parent@michigan.gov**



STATE OF MICHIGAN

JENNIFER M. GRANHOLM
GOVERNOR

DEPARTMENT OF COMMUNITY HEALTH
LANSING

JANET OLSZEWSKI
DIRECTOR

To: Michigan Birthing Hospital Coordinators
From: State Newborn Screening
Re: Changes in the First and Repeat newborn screening cards

New/changed fields on **First** cards are:

ANTIBIOTICS?
ANY RBC TRANSFUSION? (Previously RBC TRANSFUSION?)
ANY TPN FEEDING? (Previously TPN FEEDING?)
ARAB DESCENT (replaces MIDDLE EASTERN)
MOM/BABY STEROID TX?
PHYSICIAN FAX

New/changed fields on **Repeat** cards are:

INFANT'S AGE (hrs) - **this field NOT to be filled out***
INFANT'S BIRTH WEIGHT (gms) - **this field NOT to be filled out***
ANTIBIOTICS?
ANY RBC TRANSFUSION? (Previously RBC TRANSFUSION?)
ANY TPN FEEDING? (Previously TPN FEEDING?)
ARAB DESCENT (replaces MIDDLE EASTERN)
BABY STEROID TX?
PHYSICIAN FAX

On both the First and Repeat cards for

1. ANY TPN FEEDING?

Please answer YES if baby on TPN feeding at any time not just at the time of the current specimen draw. If marked Yes, and a scientist needs dates, a call will be made to obtain this information.

2. ANTIBIOTICS?

Please answer YES if baby on Antibiotics at any time not just at the time of the current specimen draw. If marked Yes, and a scientist needs dates, a call will be made to obtain this information.

3. MOM/BABY STEROID TX?

Please answer YES if mom or baby on Steroid Tx at any time not just at the time of the current specimen draw. If marked Yes, and a scientist needs dates, a call will be made to obtain this information.

On the Repeat cards for

1. BABY STEROID TX?

This appears under the MOTHER section because there was not room under BABY section. Please answer YES if baby on Steroid Tx at any time not just at the time of the current specimen draw. If marked Yes, and a scientist needs dates, a call will be made to obtain this information.

***Manufacturer could not delete the field as requested**

BABY

LAST NAME FIRST NAME BIRTH DATE BIRTH TIME (Military) BIRTH WT. (gms) WKS GESTATION GENDER

SPECIMEN DATE COLLECTION TIME (Military) Collected By: (Initials)

MEDICAL RECORD #

SINGLE BIRTH MULTIPLE BIRTH →

ANY RBC TRANSFUSION? YES DATE

NICU/ SPECIAL CARE? YES NO

ANY TPN FEEDING? YES NO

HISPANIC NON-HISPANIC

WHITE AMERICAN INDIAN

BLACK ASIAN/PACIFIC ISLAND. MULTI-RACIAL

ARAB DESCENT

BIRTH ORDER A B C

ANTIBIOTICS? YES NO

MOM/ BABY STERIOD TX? YES NO

MDCH USE ONLY

MOTHER

LAST NAME FIRST NAME PHONE

ADDRESS CITY STATE ZIP

SOCIAL SECURITY NUMBER

MEDICAL RECORD #

BIRTH DATE

HEPATITIS B SURFACE ANTIGEN (HBsAg) TEST DATE RESULT POSITIVE NEGATIVE

PHYSICIAN

LAST NAME FIRST NAME PHONE FAX

SUBMITTER NAME

HOSPITAL CODE (if applicable) 00

SUBMITTER

ADDRESS CITY PHONE

BIRTH HOSPITAL (if different from submitter)

MDCH use only

MI Dept. of Comm. Hlth.
By Authority of Act 568
P.A. MCLA 333.5431



FIRST SAMPLE

Please DO NOT fill out these 2 fields on Repeat Specimen card

BABY

LAST NAME FIRST NAME GENDER ☐ MALE ☐ FEMALE BIRTH ORDER ☐ A ☐ B ☐ C ☐ D ☐ E ☐ F ☐ G ☐ H ☐ I ☐ J ☐ K ☐ L ☐ M ☐ N ☐ O ☐ P ☐ Q ☐ R ☐ S ☐ T ☐ U ☐ V ☐ W ☐ X ☐ Y ☐ Z

BIRTH DATE MONTH DAY YEAR SPECIMEN DATE MONTH DAY YEAR MEDICAL RECORD #

INFANT'S AGE (hrs) MONTH DAY YEAR INFANT'S BIRTH WEIGHT (gms) CURRENT WT. (gms) SINGLE BIRTH ☐ MULTIPLE BIRTH ☐ ANY RBC TRANSFUSION? ☐ NO ☐ YES DATE MONTH DAY YEAR NUCU/ SPECIAL CARE? ☐ NO ☐ YES ANY TPN FEEDING? ☐ NO ☐ YES ANY RBC TRANSFUSION? ☐ NO ☐ YES DATE MONTH DAY YEAR

Collected By: (initials) MONTH DAY YEAR

HISPANIC ☐ NON-HISPANIC ☐ WHITE ☐ AMERICAN INDIAN ☐ BLACK ☐ ASIAN/PACIFIC ISLAND. ☐ ARAB DESCENT ☐ MULTI-RACIAL ☐ BIRTH ORDER ANTIBIOTICS? ☐ NO ☐ YES

MDCH USE ONLY

MI Dept. of Comm. Hlth. By Authority of Act 568 P.A. MCLA 333.5431

MOTHER

LAST NAME FIRST NAME ADDRESS CITY STATE ZIP PHONE SOCIAL SECURITY NUMBER BIRTH DATE MONTH DAY YEAR

PHYSICIAN

LAST NAME FIRST NAME PHONE FAX

SUBMITTER

SUBMITTER NAME ADDRESS CITY STATE ZIP PHONE

BIRTH HOSPITAL (if different from submitter)

REPEAT SAMPLE HOSPITAL CODE (if applicable) 00

MDCH use only

NS

If you would like to receive the NBS Update, have previously requested to be placed on the mailing list, have additions, corrections or deletions, please *complete the information below* and *return this page* to the address listed below.

If preferred, you may also send an e-mail: mdch-newbornscreening@michigan.gov

Please indicate if your preference is to receive this newsletter by e-mail or regular mail.

Name _____ Title _____

Hospital or other institution _____


Address _____

E-mail address _____ Phone _____

Do you want to receive this newsletter by regular mail _____ or e-mail _____?

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PO Box 30195
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Phone: (517) 335-9205
Fax: (517) 335-9419
Email: mdch-newbornscreening@michigan.gov
Website: www.mi.gov/newbornscreening

The logo is a dark green oval with the text "Michigan Newborn Screening Program" in white, centered within it.

Michigan Newborn Screening
Program